



Duchenne  
UK

# Research Portfolio End of Year Report 2023

# Thank you Joining Jack

Thank you for your continued support and invaluable contributions to date towards our efforts to end Duchenne. The purpose of this report is to provide an end-of-year progress update on all active and completed projects\* funded by Joining Jack in the Duchenne UK portfolio, and an overview of new funding opportunities in the pipeline.

## 2023 Highlights for Joining Jack supported projects:

### Active projects:

11 active and expected to be completed between 2023 and 2026. Some projects are progressing towards original timelines however others towards newly agreed timelines. 2 projects are anticipated to start in the New Year.

### Completed Projects:

6 completed, 1 terminated based on lack of progress.

### Funding to be reallocated:

£18,882.77. This includes one project completed in 2022.

## Active Projects

### DUK-2018-27 Neuromuscular Complex Centre

**Institution: University of Newcastle**

**Principal investigator: Chiara Marini-Bettolo**

**Status: Delayed; End date of May 2023**

**Funding contribution: £10,000**

The patient and carer survey to better understand the unmet needs of patients with neuromuscular disorders to inform the development of a comprehensive neuromuscular centre model was finalised in the summer and is still being circulated to gather additional responses as initial pick-up rates were low.

We expect to receive an update by the end of year to inform of the outputs and clarify next steps.



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\*Please note that projects that have closed in 2022 and before are not included.

## **DUK-2020-08 DMD Hub Manager**

**Institution: University of Newcastle**

**Principal investigator: Emma Heslop**

**Status: On track; End date of December 2024**

**Funding contribution: £5,000**

The DMD hub continues to engage with industry, facilitating new trials in the UK and 2023 has been a fantastic year:

15 companies and 2 CROs engaged

25 open clinical trials

9 trials at feasibility stage

And 574 children have been able to access clinical trials since the DMD Hub was established in 2016.

A strategy day was held on the 11th of December 2023 to bring together the clinical network and other key stakeholders to inform next steps and priorities to continue to consolidate and expand the impact of the DMD Hub in the coming years.

## **DUK-2021-02 Hydrotherapy clinical trial**

**Institution: Lancashire Teaching Hospital**

**Principal investigator: Christian De Goede**

**Status: On track; no-cost extension awarded, end date of December 2025**

**Funding contribution: £118,000**

A no-cost extension request was awarded in April 2023 to facilitate recruitment for the hydrotherapy sessions following initial challenges. The latest update showed that recruitment expanded; Royal Manchester Children's Hospital is now open as a second site, and it is expected that Robert Jones and Agnes Hunt Hospital will be the third.

The project is progressing according to the latest agreed timeline through the experimental stage; a cohort started hydrotherapy in August 2023.

The next interim report is due by the end of this year.



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## **DUK-2021-07 Innate immune response**

**Institution: Binghamton university**

**Principal investigator: Nagaraju Kanneboyina**

**Status: On track; no-cost extension request awarded, end date of December 2024**

**Funding contribution: £1,000**

A no-cost extension request was granted in November 2023 to allow for the completion of experiments proposed in the project aims whilst publications are under review. The aims are taking longer than expected to complete, due to the complex nature of the animal models used.

We received an interim report with initial data was shared in February 2023, but the full data set will be required to draw conclusions.

## **DUK-2022-01 PhD – Antifibrotic screening platform and testing**

**Institution: University of Newcastle**

**Principal investigator: Jordi Diaz-Manera**

**Status: On track; End date of August 2025**

**Funding contribution: £32,688**

The PhD post was successfully appointed in August 2022 and substantial progress has been made during the first year; the interim report showed that the first aim of characterising the potential cytotoxic effect of the selected drugs in vitro has been completed and treatment proved safe to progress to in vivo studies in mouse models.

## **DUK-2022-02 Neuropsychiatrist post/psychosocial WS**

**Institution: Devon Partnership Trust**

**Principal investigator: Rory Conn**

**Status: On track; End date of July 2025**

**Funding contribution: £65,977.61; £9,022.39 underspent (already reallocated to DMD care UK conference 2023)**

Rory has completed his first year in post and has made great progress building relationships with colleagues across various NorthStar Sites and Satellite district general hospitals such as Oxford, Edinburgh, and Southampton.

As of this summer, 14 patients with Duchenne have been directly assessed and uptake is expected to improve in Year 2.

Monthly online surgeries are currently being run for clinicians to discuss cases relating to psychosocial care, and the prospect of a regular online 'Dad's Group' has been established, recognising the importance of supporting parents.



## **DUK-2022-08 DMD Hub Research Fellow Support (Leeds)**

**Institution: Leeds Teaching Hospital**

**Principal investigator: Anne-Marie Childs**

**Status: On track; End date of September 2024**

**Funding contribution: £35,000; £7,362.47 to be reallocated**

The clinical research fellow (Dan) funded through this award has made a significant impact, facilitating the set up and running of numerous clinical trials (including ESSENCE, MIS5ION, and Fibrogen) and supporting the PI and trial coordinators in identifying and recruiting patients, including through the DMD Central Hub Recruitment Project.

Dan is currently establishing a national research project looking at the impact of ethnicity and socio-economic status on clinical outcomes for boys with DMD in collaboration with the Northstar Network.

## **DUK-2022-10 Smart Suit**

**Status: On track**

**Funding contribution: £432,051**

The SMART Suit project has developed at a pace this year. Following the research we did with young people, families and clinicians at the end of last year, we created three concept visions for the Suit, which we presented back to these stakeholders in May. The 'Drape' concept was identified as the preferred option, and in July we set about creating a 'Monster Rig' to test our initial ideas in the lab.

In August we created our first prototype to test with young people, the learning from which we incorporated into a second prototype that was tested with young people in November. Since then, we have been busy engaging with our Technology Advisory Board and others, whose expertise covers important areas such as biomechanics, wheelchair seating and medical device regulation, to help us understand how well the current design is performing.

We are planning to build two 'Alpha' prototypes in the new year, which we will demonstrate at our New Horizons conference in early March, and which we will take to show the world at South by Southwest, an international technology and entertainment show held in Austin, Texas, that we have been selected to attend as part of the British Council contingent! Following these two shows, we plan to take the Alpha prototypes on a UK roadshow to enable the DMD and SMA communities to discuss their thoughts and expectations with us, which will inform the next phase of development.

## **DUK-2023-01 Nutrition**

**Institution: University of Glasgow**

**Principal investigator: Jarod Wong**

**Status: To start in January 2024**

**Funding contribution: £204,073**

We are pleased to share that the nutrition project will commence in January 2024.

Some aspects of the research are already underway:

- Recruitment has been successful as both a research assistant and dietician have been appointed.
- A draft of the survey has been developed
- Discussion concerning accessing data from other sites for the retrospective study
- Discussion concerning patient engagement
- Development of the case control energy balance study

## **DUK-2023-02 DMD Care UK project Manager**

**Institution: University of Newcastle**

**Principal investigator: Michela Guglieri**

**Status: On track; End date April 2024**

**Funding contribution: £22,245**

Cathy's post continues to make a significant impact through leading and developing the coordination of Newcastle University's NMD Standards of Care programme and the strategic and operational management of DMD Care UK.

## **DUK-2023-04 Aparito DMD Home Validation Study**

**Institution: Aparito**

**Principal investigator: Elin Haf-Davis**

**Status: On track; End date September 2026**

**Funding contribution: £173,336; £5,000 to be reallocated**

This project started as of October 2023 and is now making good progress following initial delays in contract approval.

The screening criteria for patients has been developed and the study team are currently moving forward with ethics submission.

Recruitment is to be carried out via the DMD hub's Clinical Trial Finder, DMD Hub Central Recruitment Database, DUK social media channels and at DUK's New Horizons conference, which will take place in March 2024.

## **DUK-2023-07 DMD Care Phase 2**

**Institution: University of Newcastle**

**Principal investigator: Michela Guglieri**

**Status: Proposal in development**

**Funding contribution: £150,000**

The scope and budget of phase 2 is currently being finalised.

# **Completed Projects**

## **DUK-2018-03 PRO instrument development**

**Institution: University of Rochester**

**Principal investigator: Chad Heatwole, Professor of Neurology**

**Status: Completed; awaiting publication**

**Funding contribution: £20,000; £270.30 to be reallocated**

This project was completed during the summer of 2022 however the first manuscript (which is currently under review) to be published and we could only complete our financial reconciliation this year.

The PRO instrument has now been fully developed and validated and can be used by companies.

## **DUK-2018-14 Taurine**

**Institution: University of Western Australia**

**Principal investigator: Peter Arthur, Associate Professor**

**Status: Completed; publications shared**

**Funding contribution: £50,000**

Unfortunately, the research carried out in the new dystrophic rat model concluded that taurine did not protect muscle from ongoing damage.

What is promising is that we have developed further evidence to support that neutrophils are a potential therapeutic target, and also identified a new blood biomarker that could be further developed to monitor disease progression in patients.



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## **DUK-2019-08 Pluripotent Stem Cells**

**Institution: University of Minnesota**

**Principal investigator: Rita Perlingiero**

**Status: Completed; publications shared**

**Funding contribution: £188,400**

This project has proved highly successful. The researchers have been able to develop a method to generate engraftable skeletal myogenic progenitors from pluripotent stem cells through conditional expression of Pax4 or Pax7 resulting in highly efficient generation of therapeutic myogenic progenitors which when transplanted into dystrophic mice locally or systemically produce large quantities of functional skeletal muscle tissue that incorporates normally into the host muscle. The success of the findings has led to IND-submission (which was expected at the end of submission) and the initiation of a Phase 1 trial in the coming months, funded through a joined DUK and PPMD grant call.

## **DUK-2019-10 Antifibrosis and Muscle Regeneration**

**Institution: University of Geneva**

**Principal investigator: Olivier Dorchies**

**Status: Terminated; existing results to be disseminated**

**Funding contribution: £25,000; £6,250 to be reallocated**

Unfortunately, in March 2023 a decision was made to terminate this project. Initially the project was severely delayed due to COVID which presented challenges relating to laboratory resources including having to change mouse models. As a result, an extension was granted however the project did not make significant progress thereafter. A final report was submitted in October 2023 and as expected, the aims were not achieved, however the findings on the new FU-5CV animal model for DMD are encouraging and should be published to ensure this model can be more widely adopted and used.

## **DUK-2019-14 Orthotics**

**Institution: Newcastle upon Tyne Hospital**

**Principal investigator: Anna Mayhew**

**Status: Completed; awaiting publications**

**Funding contribution: £10,000**

The study was completed successfully in June. The findings demonstrated that compliance to stretches was high across the sample and participants were highly satisfied with the CCD device and service.

There were difficulties in recruitment initially due to COVID and R&D approvals and only 5 participants were enrolled over the study period.

Whilst recruitment has played a role in not being able to estimate the sample size required to power a larger study, the project has successfully assessed feasibility and trial design and has identified areas for further investigation with perhaps the most significant learning point being the potential significance of adherence and its impact on outcome.



## **DUK-2020-07 Immunology project**

**Institution: University of Florida**

**Principal investigator: Manuela Corti**

**Status: Completed; awaiting publications**

**Funding contribution: £185,000**

This project was successfully completed in September and a manuscript has been submitted to Human Gene Therapy in November.

The study demonstrated a significant advancement toward the development of immunosuppressive modalities to enhance microdystrophin expression, even in the presence of pre-existing immunity. The findings show great potential for improving efficacy and durability of AAV-mediated gene therapy for DMD, and we are discussing with the research team how to take this work forward.

## **DUK-2020-13 DMD Care UK**

**Institution: University of Newcastle**

**Principal investigator: Michela Guglieri**

**Status: Completed**

**Funding contribution: 65,000**

Phase 1 of DMD Care UK was completed in May 2023 and it has proved incredibly successful. Through engaging with different specialists involved in the care and management of patients with DMD, the programme has enabled increased awareness of DMD care across the UK and delivered significant impact to the community, empowering families with the knowledge of what care should look like:

- 11 Working Groups have now been established and they are in the process of convening an additional 4 groups (Palliative Care, Nutrition, Anaesthetics, Occupational therapy and wheelchair).
- Bone and endocrine and Cardiac guidelines have been published in open access journals.
- Emergency, and respiratory guidelines have been finalised.

The proposal for phase 2 is currently under review.

# Looking ahead to 2024

We are developing several exciting funding opportunities including new research focused on treating the heart, developing second generation gene therapies and other genetic treatments; our DMD patient data platform; more projects to drive better care for all through DMD Care UK.

## Thank you

We are so grateful to Joining Jack and all your supporters for your relentless drive in our shared fight to end Duchenne. We hope that this report highlights the incredible impact and progress that your funding has enabled. Duchenne UK is enormously proud to have Partner Charities like you. Together, we will end Duchenne.

